



Sirenomelia Apus – A Case Report

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Abstract

Sirenomelia (mermaid syndrome) an extreme example of caudal regression syndrome is a very rare congenital foetal anomaly which invariably presents with lower limb fusion, sacral and pelvic bony anomalies, absent external genitalia, imperforate anus, and renal agenesis or dysgenesis. There are approximately 300 cases reported in the literature. This syndrome is incompatible with life due to the association of several congenital visceral abnormalities. Our case was a live born, normally delivered at term by a 32-year-old second gravida of lower socioeconomic status with overt diabetes mellitus. Examination of the baby revealed caudal dysgenesis having fusion of lower limbs, single lower tapering web like lower extremity with no feet and absence of external genitalia and anus. The baby died minutes after birth and we report this case due to its rarity and term live birth.

Keywords: *sirenomelia, caudal regression syndrome, lower limb fusion, maternal diabetes mellitus.*

Introduction

In Greek mythology, the Sirens were three creatures with the head of a woman and the body of a bird from the wings down. Over time, these bird-women were portrayed as more aquatic creatures, and eventually with a full mermaid-like appearance.

Sirenomelia is a rare and fatal congenital defect characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacro-coccygeal agenesis, genitourinary, and anorectal atresia. However, sirenomelia is also associated

with malformations of the upper part of the body, including cleft palate, upper thoracic and cervical vertebra abnormalities, pulmonary hypoplasia, and cardiac defects. The first medical description of Sirenomelia was by Rocheus and Polfyr way back in the sixteenth century. Duhamel in 1961 defined all the anomalies of mermaid syndrome and described it as the most severe form of caudal regression syndrome.

The incidence of sirenomelia is 0.8-1 case/100,000 births with male to female ratio being 3:1.

The rarity of the case is obvious from the fact that

many a gynaecologist might not have come across a case of sirenomelia in their whole professional carrier. There is a strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of foetuses with this anomaly will have mothers with diabetes.

Oligohydramnios secondary to severe renal dysplasia is universal. Maternal drug use, Rheumatoid factor, maternal diabetes are identified as the common factors. It may result from a defect, at the late gastrula stage, in the formation or remodelling of the early embryonic vasculature or in the generation of the mesodermal precursors during the third gestational week. We report a case of sirenomelia where maternal overt diabetes may have been the cause of this rare anomaly.

Case report

A 32-year-old diabetic with an unsupervised pregnancy of 38 weeks admitted with complaints of abdominal pain in Regional institute of medical sciences, Imphal. History of the patient tells that she was gravida 2 and had previous history of one abortion. She had no history of prior antenatal care and belonged to a tribal community with lower socioeconomic status. There is no history of exposure to tobacco both before and during the pregnancy. There was no declared exposure to other drugs. She was otherwise healthy with no known history of genetic or congenital anomaly in her family. Ultrasonography of the case revealed a live fetus, oligohydramnios and renal agenesis, severe caudal malformation, including fused lower extremities. The detailed evaluation was not possible because of the presence of severe oligohydramnios. The pictures of ultrasound were not included in this case report.

She delivered a term 2.5 kg baby with multiple congenital anomalies. The Apgar score was 3 at 1' and 0 at 5 min. The baby died within 20 min post birth in spite of resuscitation attempts by neonatologist.



Figure 1



Figure 2

On physical examination, the infant showed narrow chest, bilateral hypoplastic thumb and a single lower tapering web like lower extremity with no feet and absence of external genitalia and anus, umbilical cord with single umbilical artery (figure 1). There were also prominent epicanthal folds, hypertelorism, downward curved nose, receding chin, low-set soft dysplastic ears and small slit-like mouth suggestive of Potter's facies (Figure 2). Autopsy was declined by the parents. Intrapartum and the postpartum period of mother was uneventful.

Discussion

Sirenomelia is a rare congenital anomaly, till date approximately 300 cases have been reported in the literature of which 14 are from India. Most of these newborns were still born or died immediately after birth; death is usually due to renal agenesis, which is incompatible with life. Swader reported the first surviving infant in 1989. Till 2006, 6 cases of surviving infants with

mermaid syndrome were reported. The aetiology and pathogenesis of this malformation is unknown. Duhamel in 1961 stated that Sirenomelia and anorectal malformations represent the two extremes of a single comprehensive syndrome arising from an embryonal defect in the formation of the caudal region. He called it the Syndrome of Caudal Regression.¹

Anomalies observed in sirenomelia are described as the most severe form of caudal regression syndrome. Fusion of lower extremities, presence of single umbilical and persistent vitelline artery are major features of sirenomelia. Although the primary molecular defect resulting in sirenomelia is unclear, two main pathologic hypotheses namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed. According to vascular steal hypothesis, fusion of the limbs results from a deficient blood flow and nutrient supply to the caudal mesoderm, which in turn results in agenesis of midline structures and subsequent abnormal approximation of both lower limb fields.⁴ However in defective blastogenesis hypothesis, the primary defect in development of caudal mesoderm is attributed to a teratogenic event during the gastrulation stage.⁵ Such defect interferes with the formation of notochord, resulting in abnormal development of caudal structures. Maternal diabetes, tobacco use, retinoic acid and heavy metal exposure are possible environmental factors.

Maternal diabetes is considered as a causative environmental factor for CD because 10–15% of affected children have diabetic mothers (Assimakopoulos et al., 2004; Castori et al., 2010).⁶

In antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high resolution or colour Doppler sonography.³ The condition is usually incompatible with life due visceral abnormalities especially that of renal system. Exceptional cases without renal agenesis have survived, the best example being Tiffany Yorks, a 13-year-old girl who was born with fused legs.

Over the years, she has undergone numerous operations to separate her lower extremities.

The facial abnormality usually found in sirenomeliac infants known as Potter's facies, which includes large, low-set ears, prominent epicanthic fold, hypertelorism, flat nose and receding chin. When features of Potter's facies are combined with oligamnios and pulmonary hypoplasia it is known as Potter's syndrome.

Stevenson et al, dissected the abdominal vasculature of 11 cases of sirenomelia and demonstrated a pattern of vascular abnormality that explains the defect usually found in this condition.² They demonstrated a single large artery (steal vessel) arising from the high intraabdominal cavity which diverts nutrients from the caudal end of embryo. There is a strong association between this syndrome and maternal diabetes; up to 22 % of fetuses with this anomaly are known to have diabetic mothers. Fifteen percent of patients with sirenomelia have associated twinning, which is most often monozygotic.⁶

Sirenomelia has been classified into three types according to the number of lower limb bones present:

1. Sirenomelia apus: No feet only one tibia and one femur.
2. Sirenomelia unipus: One foot, two femurs two tibia, and two fibula.
3. Sirenomelia dipus: Two feet and two fused legs giving the appearance of a flipper.

Sirenomelia dipus has the most favourable outcome.⁸ Survival of children with sirenomelia depends on the associated visceral anomalies, especially renal function, rather than the sirenomelia itself. Initial treatment of these newborns includes supportive care and diverting colostomy, later management of these infants includes a multidisciplinary surgical approach involving various specialties.

Conclusion

Sirenomelia is a rare fatal congenital malformation with severe visceral anomalies that

decide the survival. Fusion of the lower limbs, which is very obvious, is less fatal. Few surviving patients need a multidisciplinary approach of treatment. When diagnosed antenatally, termination should be offered.⁷ However, prevention is possible and should be the goal. Regular antenatal checkup with optimum maternal blood glucose level in preconceptional period and in first trimester should be maintained to prevent this anomaly.

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